



COLQ gene

collagen like tail subunit of asymmetric acetylcholinesterase

Normal Function

The COLQ gene provides instructions for making a protein that plays an important role in the neuromuscular junction. The neuromuscular junction is the area between the ends of nerve cells and muscle cells where signals are relayed to trigger muscle movement.

The ColQ protein anchors another protein called acetylcholinesterase to the muscle cell membrane at the neuromuscular junction. The ColQ protein is made up of three identical parts (subunits). Each subunit attaches (binds) to a bundle of four acetylcholinesterase proteins. Acetylcholinesterase plays a role in regulating the length of signaling between nerve cells and muscle cells by breaking down the signaling protein acetylcholine.

Health Conditions Related to Genetic Changes

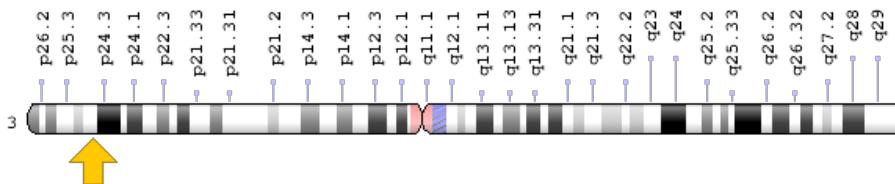
congenital myasthenic syndrome

More than 35 mutations in the COLQ gene have been found to cause congenital myasthenic syndrome. Most of these mutations change single protein building blocks (amino acids) in the ColQ protein or lead to the production of a shortened, nonfunctional protein. A lack of functional ColQ protein leads to a reduction in the amount of acetylcholinesterase that is available in the neuromuscular junction. As a result, acetylcholine is not broken down so signaling between nerve and muscle cells is prolonged. This signaling overload can damage muscle cells, leading to the muscle weakness characteristic of congenital myasthenic syndrome.

Chromosomal Location

Cytogenetic Location: 3p25.1, which is the short (p) arm of chromosome 3 at position 25.1

Molecular Location: base pairs 15,450,133 to 15,521,751 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- acetylcholinesterase-associated collagen
- acetylcholinesterase collagenic tail peptide
- AChE Q subunit
- collagen-like tail subunit (single strand of homotrimer) of asymmetric acetylcholinesterase
- collagenic tail of endplate acetylcholinesterase
- COLQ_HUMAN

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Acetylcholine and the Neuromuscular Junction
<https://www.ncbi.nlm.nih.gov/books/NBK21521/#A6207>
- Washington University, St. Louis: Neuromuscular Disease Center
<http://neuromuscular.wustl.edu/synmg.html#ache>

GeneReviews

- Congenital Myasthenic Syndromes
<https://www.ncbi.nlm.nih.gov/books/NBK1168>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28COLQ%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>

OMIM

- COLLAGENIC TAIL OF ENDPLATE ACETYLCHOLINESTERASE
<http://omim.org/entry/603033>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=COLQ%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2226
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8292>
- UniProt
<http://www.uniprot.org/uniprot/Q9Y215>

Sources for This Summary

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